

A NEW NIH STUDY FOR WOMEN AND GIRLS WITH TURNER SYNDROME

This new study aims for an in depth and comprehensive characterization of clinical features of Turner syndrome and identification of the specific genes linked to each feature. This approach is designed to benefit those with Turner syndrome by improved diagnosis and recognition of potential and existing medical problems, resulting in improved preventative and therapeutic medical care. Adult women with Turner's have a significantly higher risk for developing high blood pressure, diabetes, and osteoporosis than other women. We hope to identify the specific genes that cause many of these medical problems. By learning more about the genes that cause this disorder, we hope to improve the diagnosis and treatment of girls and women with Turner syndrome in the future.

One of the most distressing aspects of this disorder for many patients has been the lack of medical and scientific information on the diagnosis and its implications. This protocol will include genetic counseling and will provide TS participants with opportunities to ask questions and discuss their diagnosis and medical/social issues with the physicians, research nurses and genetic counselors involved in the study. Indeed, a specific goal of the study is the identification of major themes of interest and concern to people affected with Turner syndrome and development of a counseling program that will effectively address these concerns. The results of clinical and genetic studies will also be provided in written form to study subjects and to their physicians.

This study requires a 4-5 day inpatient stay at the NIH hospital in Bethesda, MD.

Testing includes:

- ◆ Physical exam
- ◆ Body measurements
- ◆ Blood and urine sample collections
- ◆ Thorough evaluation of the heart, kidneys, and bones, including: x-rays of the wrists, MRI , ultrasounds, EKG, DEXA scan, CT scan [adults only], echocardiogram, and blood pressure monitoring
- ◆ Brain function and learning ability evaluations
- ◆ Psychosocial evaluations
- ◆ Genetic counseling and needs assessment
- ◆ DNA analysis

Study investigators will discuss results of testing with participants in a follow-up outpatient visit or over the phone. Participants will be given a written summary of all results. The hospital stay and testing are free to all participants.

You may take part in this study if:

- You have been diagnosed with Turner syndrome
- You are 7 years of age or older
- Your thyroid function tests are normal (or presently corrected by treatment)
- You are not pregnant

For more information please contact **Lori Hanton, RN, Study Coordinator,**
at **1-888-437-4338** or **(301) 496-0862** if in the Washington, D.C. area.